TX RARE

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Committee on Energy & Commerce
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Negotiating a Better Deal: Legislation to Lower the Cost of Prescription Drugs

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Chairwoman Eshoo, Ranking Member Guthrie, and distinguished members of the Health Subcommittee of the House Committee on Energy and Commerce. I am privileged to be here today as a rare disease parent, caregiver, and patient advocate to share my perspective, and represent the 1 in 10 Americans affected by more than 7,000 rare diseases. I am the Founding President of Texas Rare Alliance, an education and advocacy organization working to improve access and health outcomes for nearly 3 million Texans living with a rare disease. That is a large number, and it's correct. There are more Americans living with a rare disease than have HIV, heart disease, or stroke combined. Only 5 percent have an FDA-approved treatment. It is crucial we continue research and development of additional rare disease treatments, and that rare disease patients can access disease-modifying treatments upon approval.

We know what price controls have done to patients in other countries – they have resulted in patients, including children like Hunter, having worse access to treatments. They undervalue the lives of people with rare diseases, and disabled people.

I. Hunter's Diagnostic Odyssey

On July 31, 2001, our newborn son, Hunter, was taken to the NICU at birth and intubated for respiratory failure. The doctors believed his lungs were underdeveloped, but his carbon dioxide levels continued to rise, and he faced organ failure. The doctors believed something else was wrong, but they didn't know what it was.

Fortunately, Hunter was successfully extubated, and came home after 11 days in the NICU. We thought everything would be okay, but we were mistaken.

Hunter lost nearly all movement at two weeks of age. We didn't know what was happening. *At one point my husband asked me if I had shaken Hunter.* I could never hurt our baby, but he was hurting. The coming days and weeks led to more heartache and questions, but no answers.

On September 30, 2011, our world changed forever. Doctors diagnosed our eight-week-old baby with Spinal Muscular Atrophy (SMA). SMA is like ALS in babies; it robs the ability to move, swallow, and ultimately breathe, and is the number one genetic cause of death for babies.

Doctors told us there was no treatment and no hope. They instructed us to take Hunter home and enjoy what time we had left with him, which they expected to be about three months.

II. Hunter's Pathway to Treatment

We could not afford to follow the advice of the doctors who diagnosed Hunter. The stakes were just too high. We had to try to save our sweet baby.

Desperation drove our relentless pursuit of a treatment.

A researcher answered our plea and agreed to help. He provided us with the chemistry to a compound. We manufactured that compound in the US and took it to Mexico for an N of 1 trial.

And so, Hunter's reverse Dallas Buyer's Club adventure began.

He received his first lifesaving treatment just 8 weeks after his diagnosis-the day before Thanksgiving-making him the first SMA patient to receive a lifesaving treatment.

Hunter continued his treatments.

That left me unable to reconcile how quickly we secured a lifesaving treatment for Hunter with how long it takes to bring treatments to desperate patients with life threatening conditions. I knew by experience we could do much better, and I began advocating to improve access to the diagnosis and treatments.

My goal is to help secure presymptomatic diagnosis and treatments for all rare disease patients. In March of 2016, I was asked to attend an FDA meeting as a core member of the Families for Acceleration of SMA Treatments (FAST) on May 4, 2016 where we advocated for early access to the first SMA treatment on the grounds that real world evidence established the primary endpoint had been met, leading the FDA to approve Spinraza (nusinersen), the first treatment for SMA, on December 23, 2016, for pediatric and adult SMA patients[1] making it the fastest FDA approval ever.

Like many other SMA infants and children, Hunter participated in the Spinraza EAP. He received his first Spinraza treatment on November 21, 2016, just 2 days shy of the 5-year anniversary of his first treatment in Mexico.

III. The Unmet Need for Rare Disease Treatments

The rare disease landscape in the United States, while improving, has a long way to go. 1 in 10 Americans, over 32 million, has a rare disease.[2] Half of rare disease patients are children (16 million

[1] U.S. Food & Drug Admin. (Dec. 23, 2016). *FDA Approves First Drug for Spinal Muscular Atrophy* [FDA News Release]. Retrieved from https://www.fda.gov/news-events/press-announcements/fda-approves-first-drug-spinal-muscular-atrophy
[2] *RARE Facts* (n.d.). Global Genes. https://globalgenes.org/rare-facts/

American children).[3] 35% of the deaths in the first year of life are caused by rare diseases.[4] 30% of children with a rare disease will not survive to their 5th birthday.[5] There are 7,000.[6]

95% of all rare diseases do not have an FDA-approved disease-modifying treatment.[7] It will take thousands of years to realize treatments for all rare diseases if FDA-approved therapies continue at the current rate.[8]

I am intimately aware of what it means to lack an approved rare disease treatment for your rare disease community. It is devastating. We need more conditions like SMA to cross over from the 95% percent of rare diseases that lack an approved SMA treatment to the 5% with an FDA-approved disease modifying treatment. To achieve this, we must encourage continued research and development of breakthrough therapeutics.

IV. Understanding the True Economic Burden of Rare Diseases

The National Economic Burden of Rare Disease Study in the U.S. published by the EveryLife Foundation for Rare Disease covered 379 rare diseases affecting 15.5 million people in the U.S. for 2019. The study estimated the overall rare disease economic burden to exceed $966 billion.[9] This included "$418 billion in direct medical cost and $548 billion in indirect and non-medical costs absorbed directly by families living with rare diseases."[10]

Accordingly, in-direct and Non-medical costs (costs absorbed directly by families) in 2019 accounted for nearly 60% of the overall cost.[11] **Prescription medications and outpatient prescription administration were only about 10% of the overall economic burden and less than what was spent on inpatient care.[12] We can't expect to address affordability if we are focusing on a small percentage of the problem.**

V. Protecting the Pathway to Rare Disease Research

Congress recognized the unmet need for rare disease treatments by passing the Orphan Drug Act (ODA) in 1983.[13] The ODA incentivizes the development of treatments for rare diseases, many of which are life-threatening, and most lack an approved treatment.[14] Congress reaffirmed its commitment to the rare disease community in 2016 by passing the 21st Century Cures Act,[15] a

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[3] *Id.*
[4] *Get the Facts on Rare Diseases* (n.d.). Rare Genomics Institute. https://www.raregenomics.org/rare-disease-facts
[5] *Id.*
[6] *RARE Facts* (n.d.). Global Genes. https://globalgenes.org/rare-facts/
[7] *Id.*
[8] Bock, Eric. (Apr. 17, 2020). Rare Disease Research Progressing, But Could Go Even Faster. *NIH Record.* 72(8) 1, 8-9. https://nihrecord.nih.gov/sites/recordNIH/files/pdf/2020/NIH-Record-2020-04-17.pdf
[9] EveryLife Foundation for Rare Diseases. (2021). *The National Economic Burden of Rare Disease Study*. 15 https://everylifefoundation.org/wp-content/uploads/2021/02/The_National_Economic_Burden_of_Rare_Disease_Study_Summary_Report_February_2021.pdf
[10] *Id.*
[11] *Id.*
[12] *Id.*
[13] Orphan Drug Act of 1983. Pub L. No. 97–414, 96 Stat. 2049.
[14] Rare Diseases at FDA. (Feb 20, 2020) US Food and Drug Administration. https://www.fda.gov/patients/rare-diseases-fda

[15] Public Law 114–255, 130 STAT. 1033. To Accelerate the Discovery, Development, and Delivery of 21st Century Cures, and for Other Purposes. Enacted: December 13, 2016. https://www.congress.gov/114/plaws/publ255/PLAW-114publ255.pdf

bipartisan effort President Obama signed into law. The Act included many provisions to improve the discovery, development, and delivery of orphan therapies for rare disease patients, together with substantial NIH funding.[16]

VI. The Commitment to Rare Disease Research Is Paying Off

2018 represented a historic year for the rare disease community. For the first time, rare disease approvals exceeded general approvals from the Center for Drug Evaluation and Research (CDER) at the FDA.[17] "In 2018, 34 of CDER's 59 novel drugs (58%) were approved to treat rare or "orphan" diseases that affect 200,000 or fewer Americans."[18] Increased approvals of rare disease treatments is a welcome sign for the rare disease community. Future policies should avoid barriers to research and development as we continue efforts needed to secure FDA-approved therapies for rare diseases with unmet needs.

VII. Rare Disease Research Benefits Us All

Research for rare diseases greased the wheels for a COVID-19 vaccine. Prior to the Covid-19 pandemic, most had not heard of mRNA technologies. This is not true for researchers in the rare disease community. There are more than 145 ongoing mRNA clinical trials for rare diseases, including Cystic Fibrosis.[19] This represents more than 25% of all mRNA clinical trials.[20] Rare disease researchers pivoted to work on developing treatments and vaccines for Covid-19. One of these researchers is Dr. David Fajgenbaum, a clinician and researcher who also happens to be a rare disease patient with Castleman Disease.[21]

Today, mRNA Covid vaccines protect most vaccinated patients in the US. As of April 28th, 234.6 million vaccines have been administered in the US. 226.5 million of those vaccines-96.5%- have been the Pfizer & Moderna mRNA vaccines.[22] **We need research into breakthrough technologies like mRNA technology to continue and protect all of us.**

VIII. My Experience with QALY-Based Cost Effectiveness Analysis as is Used in Other Countries

Knowing that H.R. 3 was scored assuming use of quality-adjusted life years by the Secretary to set prices, and that foreign countries rely on the metric, it is important to understand why the patient and disability communities so adamantly oppose it. Our 9-year-old son, Hunter has a QALY of .2 because he has SMA.[23] This effectively means that his life is valued at 20% of that of our children who are in

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[16] Huron, Jennifer. President Signs 21st Century Cures Medical Innovation Bill Into Law (Dec. 13, 2016). National Organization of Rare Disorders. https://rarediseases.org/president-signs-21st-century-cures-bill-law/
Rare Diseases at FDA. (Feb 20, 2020). US Food and Drug Administration. https://www.fda.gov/patients/rare-diseases-fda
[17] Center for Drug Evaluation and Research. Advancing Health Through Innovation: 2018 New Drug Therapy Approvals. https://www.fda.gov/files/drugs/published/New-Drug-Therapy-Approvals-2018_3.pdf
[18] *Id.*
[19] CBI Insights. (Feb. 3, 2021). What Are mRNA Therapies, and How Are They Used for Vaccines? Research Briefs. https://www.cbinsights.com/research/what-are-mrna-therapies/
[20] *Id.*
[21] Moffitt, Debra. (May 27, 2020). Rare Disease Hunter Dr. David Fajgenbaum Takes on COVID-19. https://www.cslbehring.com/vita/2020/david-fajgenbaum-takes-on-covid19
[22] Fry, Erika & Rapp, Nicolas. (Apr. 28, 2021). 55% of U.S. Adults Have Gotten a COVID Vaccine. See How Your State Is Doing. Fortune. https://fortune.com/2021/04/28/covid-vaccine-tracker-update-us-state-by-state-pfizer-moderna-johnson-and-johnson-data-coronavirus-vaccines-april-2021/
[23] QALY Scale. https://valueourhealth.org

good health. I understand the ramifications of QALYs in other countries, and the thought of the U.S. discriminating against Hunter's life that would result in decreased access to medications and medical services that he needs to survive is beyond devastating. ALS is another example worth noting, because it has a QALY of -.05.[24] This tells ALS patients and their loved ones that having ALS is worse than being dead.

We are blessed that Hunter hasn't been directly impacted by the QALY. However, I did move the family back to our St. Louis home last summer and fall following the death of Michael Hickson in Austin by a doctor who employed QALY rationale to deny treatment because the doctor didn't perceive Michael as having a quality of life.

It was a sucker punch. I packed up everything and ran away fast back to St. Louis where I knew St. Louis Children's Hospital saw value in Hunter, and worked hard to save him so many times, and had provided his Spinraza treatments.

I also witnessed Hunter's friend Ben impacted by QALYs. Ben and Hunter started the Spinraza EAP together. Under the EAP, Biogen provided Spinraza, and St. Louis Children's Hospital waived fees for administration. Upon FDA-approval, insurers were to cover Spinraza and the costs of administration. United drug its feet in developing its coverage for Spinraza. Biogen secured FDA approval to provide Spinraza gap treatments until insurerance policies on Spinraza became active.

Both Hunter and Ben were covered by United Choice Plus policies. Hunter met the inclusion criteria, because he only uses bipap, a machine similar to a cpap that breathes for him, at night. However, because Ben was bipap dependent, he failed to meet the inclusion criteria for coverage by United.

Ben's mom, Melissa, cried. She asked why Ben's life wasn't worth saving too. I cried with Melissa. I assured her that Ben's life was worth saving just as Hunters was. Unfortunately, what I hold to be true didn't matter, because I couldn't change the policy to cover Ben's Spinraza treatments.

Fortunately, Biogen placed Ben on a patient assistance program providing him Spinraza for his injections at no cost. Although United would not cover Spinraza, it did cover the cost of administration. Biogen continued to cover Ben in its patient assistance program until Ben secured a Missouri Medicaid Waiver which covered Spinraza.

This should provide some context for why I oppose H.R. 3. I understand the impact H.R. 3 would have on rare disease patients like Ben by importing QALYs incorporated in foreign reference pricing. We cannot import reference prices without importing reliance on QALYs utilized to determine those prices. This means children like Ben would not be able to secure access to the treatments they need to survive.

In the absence of treatments, rare disease parents work hard to keep our children alive. We are more than parents. We are forced to become medical experts providing a standard of care at home that exceeds care provided by hospitals. That's not a smug statement. When our children are in the hospital, we don't leave their side. We know the standard of care for their rare disease, and we know that if the

[24] *Id.*

hospital follows the protocol for a child of typical health, our children would be harmed, and might not survive.

We manage the machines that feed, breathe for, and monitor our babies and children. We give them their medicines and do their respiratory treatments. Sometimes we get so tired, and yet we can't take time off, because the rare diseases our children fight against never take time off.

The doctors tell us there's no hope, but we have more than hope. We have unconditional love for our children, and we refuse to give up on them. We value them. We value every breath they take, and we dare not take a single breath for granted.

Our advocacy against H.R. 3, will not stop. We cannot afford for it to stop. We know the consequences will exact a heavy toll against our children with rare disease. We refuse to save our children only to have a system adopt QALYs that gives up on them.

H.R. 3 would also greatly reduce research and development of rare disease treatments. That research and development is the stuff dreams are made of. We follow the science for our children's conditions. We hold bake sales, runs, parties, and pretty much anything we can think of to raise funds to get research started. The thing is our funds only get researchers so far. Without follow-on funding from the NIH, biotech companies, or biopharmaceutical companies, the research stalls.

Research would stall under the price controls tied to reference pricing under H.R. 3. We know it will, because we don't see clinical trials taking place in or approvals coming from those countries. They are innovation deserts. Just as deserts are a cruel place when you need water to survive, innovation deserts are relentless when you need access to a rare disease treatment to save your child.

With 95% of 7000 rare diseases lacking an approved treatment, we can't afford to turn our back to innovation. At the current pace, it will take thousands of years to secure treatments for all rare diseases. Thousands of years would be too late for any of us. A third of children with a rare disease will not survive to their 5th birthday. Rare diseases operate fast. We need nimble research labs with the capabilities and the dedication to work on multiple treatments in parallel.

Our dream for the rare disease community is that research for rare diseases will move with the same relentless urgency as Covid-19 research did. Can you imagine if rare diseases were contagious? What would happen if you quickly lost the ability to move, swallow, and ultimately breathe? That might sound like SMA, Hunter's rare disease. You might be thinking well, SMA has a treatment, and you're right. However, SMA with Respiratory Distress (SMARD) doesn't have a treatment. Like SMA, it can rob the ability to move. SMARD has one more strike against it as a rare disease—it's an ultra-rare disease. It's harder for ultra-rare diseases to provide the seed funding for researchers to begin research, because there's less families to work together and raise the funds. It's also harder to attract follow-on funding to continue the research, because with so few patients, biotech and biopharmaceutical companies are unlikely to continue the research.

Yes, SMA children like Hunter have an approved treatment, but SMARD patients don't, and they need one too.

IX. The ICER Assessment of Spinal Muscular Atrophy

We know different people respond differently to the same drugs. For many conditions, such disparities are reflected in clinical knowledge – but not yet in research literature. The metric that H.R. 3 relies on to set prices is incapable of addressing that problem. When the Institute for Clinical and Economic Review (ICER) conducted its value assessment of Spinraza, I testified that Hunter had not been hospitalized since starting treatment in 2016. It was so stressful in 2015 when Hunter was intubated, and doctors continually prepped us for the decision to trach Hunter or allow him to die if they could not successfully extubate him. Fortunately, we did not have to make that decision, but many SMA families have. This is why access to efficacious treatments that improve respiratory functions are so important. It is also imperative to commence treatments presymptomatically to afford the patients the best possible health outcomes and quality of life. I testified to ICER about all of the costs of SMA that no one considers, such as the daily respiratory protocol such as albuterol nebulizer treatment, the extensive daily stretching physical therapy to preserve and improve movement, occupational therapy to improve movements related to education and navigating the world, and speech including oral stimulation to improve both speech and swallow, not to mention nutrition and medical equipment, most of which is not covered by insurance. Yet, their assessment using a QALY measure failed to capture any of the significant economic aspects of that drug for my family. ICER ultimately determined that Spinraza was not cost effective.

In 2018, when CVS Caremark threatened to use a QALY-based benchmark for what drugs they would cover in a benefit package being marketed to their employer clients, it created a moment of panic for people that rely on these innovations to survive. Thankfully, advocacy worked against CVS Caremark and they stopped marketing that benefit package, but we live in constant fear of payers using these cost-per-QALY benchmarks to determine what they will or will not cover. I cannot imagine the fate of families like mine if we were to explicitly endorse use of discriminatory metrics in Medicare and Medicaid.

X. Setting Prices Based on a One-Size-Fits-All Reference or Metric Would be Disastrous for Access to Medications.

According to the Galen institute, 89% of new medicines introduced between 2011 and 2018 are available in the U.S. compared to 62% in Germany, 60% in the U.K., 50% in Japan, and 48% in France.[25] In its analysis of H.R. 3, the CBO estimated that the resulting reduced revenues over a decade between "$.05 trillion to $1 trillion would lead to a reduction of 8 to 15 new drugs coming to market. It is difficult to know in advance the nature of these drugs or to quantify the effect of foregone innovation on health."[26] The CBO estimated the cost of creating and maintaining the system to implement H.R. 3 would increase spending at around $3 billion over a 6-year period.[27]

XI. National Council on Disability Statements Opposing QALYs and Reference to Foreign Countries

[25] The Editorial Board. (Oct. 4, 2019). Pelosi's Expensive Drug Bill. The Wall Street Journal. https://www.wsj.com/articles/pelosis-expensive-drug-bill-11570228189
[26] Swagel, Phillip. (Oct. 11, 2019). Effects of Drug Price Negotiation Stemming From Title 1 of H.R. 3, the Lower Drug Costs Now Act of 2019, on Spending and Revenues Related to Part D of Medicare. CBO https://www.cbo.gov/system/files/2019-10/hr3ltr.pdf
[27] *Id.*

You have likely read the letter opposing H.R. 3 sent to the committee by the National Council on Disability, an independent federal agency advising Congress and the administration on disability issues.[28] The concerns from this nonpartisan agency about the metric called quality-adjusted life years or QALYs are not new and have been long-held concerns on both sides of the aisle. Because people with disabilities, seniors, and patients with chronic conditions may experience a potential for health that is less than their "healthier" counterparts, treatment that extends or improves their life may result in fewer QALYs than a treatment developed for a non-disabled or younger population where the treatment is able to return the patient to so-called perfect health. As QALYs are assigned by both quality as well as quantity of life, an incremental QALY assessment would prioritize providing treatment to a non-disabled population with a longer theoretical life expectancy, and otherwise perfect health, over a population with a disability or chronic condition.[29]

Based on the NCD's report in 2019, we know that simplified value assessments do not account for the complex experience of patients with rare diseases that are not well-represented in the research literature, particularly for communities of color. QALYS do not to take into account clinical expertise on rare disorders that may not have an extensive research literature available for use.[30] QALYs often rely on research that does not adequately account for the ways in which many people— especially, though not exclusively, those with rare conditions— may have medication responses that vary dramatically from the average.[31] For individuals with rare conditions or who come from groups underrepresented in research, like people with disabilities and people of color, the inability of QALYs to account for information that primarily exists within clinical knowledge but has not yet made it into the research literature constitutes a serious problem.[32] Many cancer drugs are not considered valuable enough to cover in the U.K. due to their use of QALYs.[33] Historically, the United Kingdom has used QALYs to justify not covering the limited drugs available for Alzheimer's disease, even when they cost the equivalent of a cup of coffee.[34] The use of a cost-per-QALY analysis in the United Kingdom delayed access to cystic fibrosis treatments in the United Kingdom.[35]

XII. QALYs are Not Ethical

America's sense of morality and ethic of equality makes it a bridge too far to deny or devalue care to those with significant lifetime health needs just because they may never achieve a pre-conceived notion of optimal health. The implications are even more significant for communities of color that are underrepresented in the research literature that informs value assessment.[36] We have dealt directly with this kind of discrimination in Texas related to COVID-19 care, and thankfully with the help of the HHS Office for Civil Rights, states are now clearly prohibited from using a patient's long-term life expectancy as a factor in the allocation and re-allocation of scarce medical resources.[37] It makes no

[28] National Council on Disability, Tletter to House Committees with concerns regarding H.R. 3 (April 29, 2021) at https://ncd.gov/publications/2021/ncd-letter-house-committees-concerns-regarding-hr-3
[29] National Council on Disability. Quality-adjusted life years and the devaluation of life with disability. (2019) at https://ncd.gov/sites/default/files/NCD_Quality_Adjusted_Life_Report_508.pdf.
[30] *Id* at p.12.
[31] *Id* at p.36.
[32] *Id* at p.37.
[33] *Id* at p.50.
[34] *Id*.
[35] *Id* at p.52.
[36] National Minority Quality Forum et al, Traditional Value Assessment Methods Fail Communities of Color and Exacerbate Health Inequities White Paper (September 28, 2020) at https://www.nmqf.org/nmqf-media/traditional-value-assessment-methods.
[37] U.S. Department of Health and Human Services, Office for Civil Rights, Revised Crisis Standards of Care for North Texas Mass Critical Care Guidelines Task Force and the Southwest Texas Regional Advisory Council, et al (January 14, 2021) at

sense then that we would allow for use of a metric that so similarly devalues older adults and people with disabilities, chronic conditions and rare diseases in determining how to pay for, and thereby access, treatments in Medicare.

The committee should understand that we did not have the National Council on Disability report explaining and opposing the QALY when H.R. 3 was first up for debate. Their report was issued later in 2019.[38] Therefore, I hope Congress will avoid policies that reference QALYs, whether from the Institute for Clinical and Economic Review (ICER) that not only uses the QALY but calls it the "gold standard,"[39] or by reference to foreign countries that rely on QALYs and similar average metrics that discriminate.

XIII. Patients Conflate Rising Out-of-Pocket Insurance Costs with Drug Costs.

According to CMS, Out-of-Pocket spending by patients grew 4.6% in 2019 to $406.5 billion comprising 11% of healthcare spending.[40] Retail prescription prices decreased in 2019 by .4%. Prescription drugs represent 10% of healthcare spending.[41] Growth in retail prescription drug spending increased 5.7% but was attributed to growth in the use of prescriptions drugs in terms of number of prescriptions dispensed.[42] Our population is aging, and aging individuals use more prescriptions. Hospital and physician and clinical services increased more dramatically than prescription drugs in 2019 at rates of 6.2% and 4.6% respectively. This represents a combined 51% of healthcare spending.[43]

XIV. Issues the Committee Should Consider in the Future

In the future, I hope the committee will spend some time considering the importance of diagnosis. Whole-genome sequencing (WGS) can help end the diagnostic odyssey for NICU and PICU patients like Hunter while reducing healthcare spending by decreasing days in the hospital, reducing unnecessary (often invasive) tests, and improving health outcomes.[44] In Texas, I proposed Project Baby Dillo (PBD) to provide WGS for low-income NICU and PICU patients in Texas to help end the diagnostic odyssey, improve health outcomes, and reduce healthcare savings. PBD was introduced by a rider and is currently under consideration for appropriations.[45]

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https://www.hhs.gov/about/news/2021/01/14/ocr-provides-technical-assistance-ensure-crisis-standards-of-care-protect-against-age-disability-discrimination.html

[38] *Id*

[39] Institute for Clinical and Economic Review, Statement on QALYs and evLYG (2017) at https://icer.org/our-approach/methods-process/cost-effectiveness-the-qaly-and-the-evlyg/

[40] National Health Expenditures 2019 Highlights (2020). CMS https://www.cms.gov/files/document/highlights.pdf

[41] *Id.*

[42] *Id.*

[43] *Id.*

[44] *Project Baby Bear Fact Sheet* (n.d.). Rady Children's Institute for Genomic Medicine. https://www.radygenomics.org/wp-content/uploads/2020/06/PBB-fact-sheet-1.pdf.

[45] Reps. Howard, Ann Johnson, Raney, and Rose. *Proposed Funding and Rider Project Baby Dillo. Rider #79-page* 83 House Committee on Appropriations Article II Riders, (Mar. 18, 2021).

An additional and essential avenue to ending the diagnostic odyssey and providing babies access to the best health outcomes possible is the passage of H.R.482 - Newborn Screening Saves Lives Reauthorization Act of 2021.[46] I also hope this committee takes up this most meaningful bill.

XV. Conclusion

I personally felt the devastation of being told there was no hope and that my newborn would die in 3 months from an imminently terminal rare disease. I've cried tears for babies and children who lost their battle with SMA and other rare diseases and watched families say goodbye. We found our way to a treatment for Hunter and helped other SMA patients access a treatment earlier. My mission is to help secure presymptomatic diagnosis and treatment for all rare disease patients.

We can improve the rare disease community by ensuring continued research and development for the 95% of rare diseases without a treatment. We must respect and value the lives of medically fragile, disabled, and elderly individuals. Their lives mater, and we cannot afford to discount their lives at any cost. With all of us working together to embrace innovation and technology, we can expedite rare disease treatments as we did with Covid-19 vaccines. We witnessed what is possible and treating rare disease patients is just as important as protecting lives from Covid-19. Please consider the negative implications that imposed drug pricing would have on the development of rare disease treatments, and access to those treatments. Thank you.

[46] Newborn Screening Saves Lives Reauthorization Act of 2021, H.R. 482, 117th Cong. (2021). https://www.congress.gov/bill/117th-congress/house-bill/482